Desmoid Tumor of the Breast: a Case Report and Review of the Literature

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ABSTRACT
Fibromatosis or desmoid tumor (DT) is a rare entity, which constitutes 0.02% of all tumors. Desmoid tumor is frequently locally aggressive with frequent recurrence even if without metastatic potential. Breast is an unusual location of this entity, and only a few cases of breast desmoid tumors have been reported. We present a case of a desmoid tumor of the left breast in a 24-years-old woman, with a review of the literature.

Keywords: fibromatosis, desmoid tumor, rare tumor, breast, recurrence

1. INTRODUCTION
Fibromatosis or Desmoid tumor is a benign slow-growing fibroblastic neoplasm, very similar to desmoid-type fibromatosis of the abdominal wall(1). With a frequency of only 0.02% of all tumors(2), this rare tumor is frequently locally aggressive, without metastatic potential(3). Breast is an unusual location; it represents 0.2% of all breast tumor types, more common among women than among men. Sporadic cases are most frequent, but desmoid tumors can be associated with familial adenomatous polyposis (FAP) or Gardner’s syndrome (GS)(4). Surgery with wide local excision is the cornerstone of the treatment. However, recurrence is common(1). Postoperative radiation is controversial, commonly used if incomplete resection. Few cases have been reported in the literature. We report a case of a desmoid tumor of the left breast.

2. CASE REPORT
A 24-year-old woman presented with a palpable left breast mass that started 1 year earlier. She did not have any other medical history. There was no family history of ovarian cancer, breast cancer, GS or FAP. She had no history of contraceptive use, and there was no previous breast surgery or trauma to the breast or chest wall. Clinical examination showed a mobile retro-nipple mass of 5 cm diameter, with normal skin and no nipple retraction.

The mammogram showed a well-defined hypoechoic lesion (52×28 mm) astride the outer quadrants in the left breast, with a cutaneous starting point, categorized as BIRAD III (Figure 1). Biopsy of the lesion demonstrated a tumor proliferation of spindle cells arranged in divergent beams. Immunohistochemical stains showed the tumor to be β-catenin and smooth muscle actin (+) and desmin and CD34 (-), that was compatible with an extra-abdominal desmoid tumor. A widened excision was performed. Pathology revealed a desmoid tumor of 5.5 cm with free surgical margins.

confirm this in their series of 33 patients with positive receptors(8).
Clinically, DT presents usually as a single palpable, unilateral, mobile, painless mass which could be fixed to the pectoralis muscle. A skin or nipple retraction may also be observed. Pain is present when deep plans are infiltrated. On imaging, typical mammograms show a high-density mass, irregularly shaped with speculated margins. On ultrasound, fibromatosis presents as an irregularly shaped, hypoechoic mass with a posterior acoustic shadowing(9). Those appearances are mimicking a breast carcinoma(10). MRI is useful to determine tumor extension and to evaluate the involvement of chest wall. It’s the best exam for the diagnosis of desmoid tumors. The lesion is typically ill-defined, heterogeneous, hypo-isointense on T1-weighted images, and hypo-hyperintense on T2-weighted images(10). On gross pathology, fibromatosis appears as a white well-circumscribed, poorly vascularized nodular mass, with firm consistency(11). Histologically, DT is characterized by a low-grade spindle cell proliferation of interlacing fibroblastic bundles, with low mitotic index and various degrees of collagen(12). On immunohistochemistry, the positive presence of actin and vimentin is very important for diagnosis. CD34 is usually negative and desmin rarely positive(13). The cytoplasmic and nuclear expression of β-catenin could be present in 70 to 80%, but it’s not specific for DT(14).
In our case, both actin and β-catenin were positives.

The management of fibromatosis is controversial. The treatment of choice is wide local excision with clear margins(15). But not all positive margins are recurrent and negative margins do not exclude the possibility of local recurrence(16,17). The chirurgical option could be avoided in many cases (negative cosmetic results) and some cases of spontaneous regression have been reported. Local recurrence rates are 21 to 27% in the literature(18). There are many factors that increase recurrence risks, such as young age, multifocality, and involved margins. This risk is higher in the first three years, which explains why breast reconstruction is not recommended during this period.

A close follow-up by MRI is recommended after DT excision. A wait-and-see approach is an option for asymptomatic tumors in a less critical site(19).

The benefit of radiotherapy is unclear. Adjuvant radiation therapy is not recommended in case of clear surgical margins. Radiotherapy can be considered in the case of incomplete resection, positive or risked resection margins, unresectable tumor, the residue after surgery or recurrence(20,21). Radiation therapy has

3. DISCUSSION

Fibromatosis or Desmoid tumors (DT) are rare tumors responsible for only 0.03% of all neoplasms and 0.2% of breast tumors(2). It has a predilection for women but can also occur in men. DT can be divided into two groups: intra-abdominal (25-35 years old, parous woman); and extra-abdominal (the frequent locations are shoulder, pelvis, thigh and chest wall)(5). Breast is an unusual location, occurring in patient aged between 15 and 60 years. Most of DT occur sporadically. Some reports suggest an association between FAP and DT, with a risk of 4 to 20% to develop this tumor(6).

The etiology is not clearly defined or understood, the more often multifactorial. A history of trauma to the chest wall, surgery or breast implants (silicone and saline) may play a role in the development of the tumor(7). Some authors have discussed the alleged role of sex hormones, and therefore a relationship between a high rate of estrogens and the development of DT. However, Devouassoux-Shisheboran et al. couldn’t
Desmoid tumor is a rare tumor, locally aggressive with a frequent recurrence that can mimic breast cancer. The confirmation of DT by IHC is necessary to give the best management. MRI is recommended to appreciate tumor extension and evaluate response to treatment. Systemic therapy and adjuvant radiation are options in patients with disease recurrence and those who cannot tolerate surgery.

4. CONCLUSION

Desmoid tumor is a rare tumor, locally aggressive with a frequent recurrence that can mimic breast cancer. The confirmation of DT by IHC is necessary to give the best management. MRI is recommended to appreciate tumor extension and evaluate response to treatment. Systemic therapy and adjuvant radiation are options in patients with disease recurrence and those who cannot tolerate surgery.

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